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Range: from begin to end ☐ Reverse complemented strand Features: ☐ SNP ☐ CDD ☑ MGC

☐ 1: NM_002335. Reports Homo sapiens low ...[gi:4505018] Links

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            Mammalia; Eutheria; Euarchontoglires; Primates; Catarrhini;
            Hominidae; Homo.
REFERENCE   1  (bases 1 to 5100)
  AUTHORS   Semenov,M., Tamai,K. and He,X.
  TITLE     SOST is a ligand for LRP5/LRP6 and a Wnt signaling inhibitor
  JOURNAL   J. Biol. Chem. 280 (29), 26770-26775 (2005)
   PUBMED   15908424
  REMARK    GeneRIF: SOST antagonizes Wnt signaling by binding to the
            extracellular domain of the Wnt coreceptors LRP5 and LRP6 and
            disrupting Wnt-induced Frizzled-LRP complex formation.
REFERENCE   2  (bases 1 to 5100)
  AUTHORS   Ai,M., Holmen,S.L., Van Hul,W., Williams,B.O. and Warman,M.L.
  TITLE     Reduced affinity to and inhibition by DKK1 form a common mechanism
            by which high bone mass-associated missense mutations in LRP5
            affect canonical Wnt signaling
  JOURNAL   Mol. Cell. Biol. 25 (12), 4946-4955 (2005)
   PUBMED   15923613
  REMARK    GeneRIF: These data suggest that HBM mutant proteins can transit to
            the cell surface in sufficient quantity to transduce Wnt signal and
            that the likely mechanism for the HBM mutations' physiologic
            effects is via reduced affinity to and inhibition by DKK1.
REFERENCE   3  (bases 1 to 5100)
  AUTHORS   Rickels,M.R., Zhang,X., Mumm,S. and Whyte,M.P.
  TITLE     Oropharyngeal skeletal disease accompanying high bone mass and
            novel LRP5 mutation
  JOURNAL   J. Bone Miner. Res. 20 (5), 878-885 (2005)
   PUBMED   15824861
  REMARK    GeneRIF: a novel LRP5 mutation may be responsible for oropharyngeal
            skeletal disease
REFERENCE   4  (bases 1 to 5100)
  AUTHORS   Hartikka,H., Makitie,O., Mannikko,M., Doria,A.S., Daneman,A.,
            Cole,W.G., Ala-Kokko,L. and Sochett,E.B.
  TITLE     Heterozygous mutations in the LDL receptor-related protein 5 (LRP5)
            gene are associated with primary osteoporosis in children
  JOURNAL   J. Bone Miner. Res. 20 (5), 783-789 (2005)
   PUBMED   15824851
  REMARK    GeneRIF: heterozygous mutations in the LRP5 gene can cause
            osteoporosis in both children and adults
REFERENCE   5  (bases 1 to 5100)
  AUTHORS   Bollerslev,J., Wilson,S.G., Dick,I.M., Islam,F.M., Ueland,T.,
            Palmer,L., Devine,A. and Prince,R.L.
  TITLE     LRP5 gene polymorphisms predict bone mass and incident fractures in
            elderly Australian women
  JOURNAL   Bone 36 (4), 599-606 (2005)
   PUBMED   15777745
  REMARK    GeneRIF: genetic variation in LRP5 seems to be of importance for
            regulation of bone mass and osteoporotic fractures
REFERENCE   6  (bases 1 to 5100)
  AUTHORS   Jiao,X., Ventruto,V., Trese,M.T., Shastry,B.S. and Hejtmancik,J.F.
  TITLE     Autosomal recessive familial exudative vitreoretinopathy is
            associated with mutations in LRP5
  JOURNAL   Am. J. Hum. Genet. 75 (5), 878-884 (2004)
   PUBMED   15346351
  REMARK    GeneRIF: Mutations in LRP5 can cause autosomal recessive or
            dominant Familial exudative vitreoretinopathy.
REFERENCE   7  (bases 1 to 5100)
  AUTHORS   Koh,J.M., Jung,M.H., Hong,J.S., Park,H.J., Chang,J.S., Shin,H.D.,
            Kim,S.Y. and Kim,G.S.
  TITLE     Association between bone mineral density and LDL receptor-related
            protein 5 gene polymorphisms in young Korean men
  JOURNAL   J. Korean Med. Sci. 19 (3), 407-412 (2004)
   PUBMED   15201508
  REMARK    GeneRIF: no association was found between a LRP5 polymorphism and
            peak bone mass in young men
REFERENCE   8  (bases 1 to 5100)
  AUTHORS   Toomes,C., Bottomley,H.M., Jackson,R.M., Towns,K.V., Scott,S.,
            Mackey,D.A., Craig,J.E., Jiang,L., Yang,Z., Trembath,R.,
            Woodruff,G., Gregory-Evans,C.Y., Gregory-Evans,K., Parker,M.J.,
            Black,G.C., Downey,L.M., Zhang,K. and Inglehearn,C.F.
  TITLE     Mutations in LRP5 or FZD4 underlie the common familial exudative
            vitreoretinopathy locus on chromosome 11q
  JOURNAL   Am. J. Hum. Genet. 74 (4), 721-730 (2004)
   PUBMED   15024691
  REMARK    GeneRIF: Familial exudative vitreoretinopathy has mutations in a
            second gene at the EVR1 locus, low-density-lipoprotein
            receptor-related protein 5 (LRP5), a Wnt coreceptor
REFERENCE   9  (bases 1 to 5100)
  AUTHORS   Hoang,B.H., Kubo,T., Healey,J.H., Sowers,R., Mazza,B., Yang,R.,
            Huvos,A.G., Meyers,P.A. and Gorlick,R.
  TITLE     Expression of LDL receptor-related protein 5 (LRP5) as a novel
            marker for disease progression in high-grade osteosarcoma
  JOURNAL   Int. J. Cancer 109 (1), 106-111 (2004)
   PUBMED   14735475
  REMARK    GeneRIF: LRP5 has a role in neoplasm metastasis
REFERENCE   10 (bases 1 to 5100)
  AUTHORS   Urano,T., Shiraki,M., Ezura,Y., Fujita,M., Sekine,E., Hoshino,S.,
            Hosoi,T., Orimo,H., Emi,M., Ouchi,Y. and Inoue,S.
  TITLE     Association of a single-nucleotide polymorphism in low-density
            lipoprotein receptor-related protein 5 gene with bone mineral
            density
  JOURNAL   J. Bone Miner. Metab. 22 (4), 341-345 (2004)
   PUBMED   15221492
  REMARK    GeneRIF: LRP5 gene is a candidate for the genetic determinants of
            bone mineral density in postmenopausal women
REFERENCE   11 (bases 1 to 5100)
  AUTHORS   Mizuguchi,T., Furuta,I., Watanabe,Y., Tsukamoto,K., Tomita,H.,
            Tsujihata,M., Ohta,T., Kishino,T., Matsumoto,N., Minakami,H.,
            Niikawa,N. and Yoshiura,K.
  TITLE     LRP5, low-density-lipoprotein-receptor-related protein 5, is a
            determinant for bone mineral density
  JOURNAL   J. Hum. Genet. 49 (2), 80-86 (2004)
   PUBMED   14727154
  REMARK    GeneRIF: LRP5 is a bone mineral density determinant and also
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            contributes to a risk of osteoporosis
REFERENCE   12  (bases 1 to 5100)
  AUTHORS   Twells,R.C., Mein,C.A., Payne,F., Veijola,R., Gilbey,M., Bright,M.,
            Timms,A., Nakagawa,Y., Snook,H., Nutland,S., Rance,H.E., Carr,P.,
            Dudbridge,F., Cordell,H.J., Cooper,J., Tuomilehto-Wolf,E.,
            Tuomilehto,J., Phillips,M., Metzker,M., Hess,J.F. and Todd,J.A.
  TITLE     Linkage and association mapping of the LRP5 locus on chromosome
            11q13 in type 1 diabetes
  JOURNAL   Hum. Genet. 113 (2), 99-105 (2003)
   PUBMED   12700977
  REMARK    GeneRIF: disease association data, largely focused in the LRP5
            region with 1,106 type 1 diabetes families, provided no further
            evidence for disease association at LRP5 or at D11S987
REFERENCE   13  (bases 1 to 5100)
  AUTHORS   Babij,P., Zhao,W., Small,C., Kharode,Y., Yaworsky,P.J.,
            Bouxsein,M.L., Reddy,P.S., Bodine,P.V., Robinson,J.A., Bhat,B.,
            Marzolf,J., Moran,R.A. and Bex,F.
  TITLE     High bone mass in mice expressing a mutant LRP5 gene
  JOURNAL   J. Bone Miner. Res. 18 (6), 960-974 (2003)
   PUBMED   12817748
  REMARK    GeneRIF: G171V transgenic mice showed an increase in actively
            mineralizing bone surface and enhanced alkaline phosphatase
            staining in osteoblasts
REFERENCE   14  (bases 1 to 5100)
  AUTHORS   Van Wesenbeeck,L., Cleiren,E., Gram,J., Beals,R.K., Benichou,O.,
            Scopelliti,D., Key,L., Renton,T., Bartels,C., Gong,Y., Warman,M.L.,
            De Vernejoul,M.C., Bollerslev,J. and Van Hul,W.
  TITLE     Six novel missense mutations in the LDL receptor-related protein 5
            (LRP5) gene in different conditions with an increased bone density
  JOURNAL   Am. J. Hum. Genet. 72 (3), 763-771 (2003)
   PUBMED   12579474
  REMARK    GeneRIF: Six novel missense mutations in this gene shown in various
            conditions with an increased bone density and bone diseases.
REFERENCE   15  (bases 1 to 5100)
  AUTHORS   Van Hul,E., Gram,J., Bollerslev,J., Van Wesenbeeck,L., Mathysen,D.,
            Andersen,P.E., Vanhoenacker,F. and Van Hul,W.
  TITLE     Localization of the gene causing autosomal dominant osteopetrosis
            type I to chromosome 11q12-13
  JOURNAL   J. Bone Miner. Res. 17 (6), 1111-1117 (2002)
   PUBMED   12054167
  REMARK    GeneRIF: localization to chromosome 11q12-13 in autosomal dominant
            osteopetrosis type I
REFERENCE   16  (bases 1 to 5100)
  AUTHORS   Boyden,L.M., Mao,J., Belsky,J., Mitzner,L., Farhi,A., Mitnick,M.A.,
            Wu,D., Insogna,K. and Lifton,R.P.
  TITLE     High bone density due to a mutation in LDL-receptor-related protein
            5
  JOURNAL   N. Engl. J. Med. 346 (20), 1513-1521 (2002)
   PUBMED   12015390
  REMARK    GeneRIF: The LRP5V171 mutation causes high bone density by
            impairing the action of a normal antagonist of the Wnt pathway and
            thus increasing Wnt signaling. These findings demonstrate the role
            of altered LRP5 function in high bone mass.
REFERENCE   17  (bases 1 to 5100)
  AUTHORS   Okubo,M., Horinishi,A., Kim,D.H., Yamamoto,T.T. and Murase,T.
  TITLE     Seven novel sequence variants in the human low density lipoprotein
            receptor related protein 5 (LRP5) gene
  JOURNAL   Hum. Mutat. 19 (2), 186 (2002)
   PUBMED   11793484
  REMARK    GeneRIF: seven novel sequence variants/polymorphisms
REFERENCE   18  (bases 1 to 5100)
  AUTHORS   Little,R.D., Carulli,J.P., Del Mastro,R.G., Dupuis,J., Osborne,M.,
            Folz,C., Manning,S.P., Swain,P.M., Zhao,S.C., Eustace,B.,
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            Tulig,C., Caruso,A., Tzellas,N., Bawa,A., Franklin,B., McGuire,S.,
            Nogues,X., Gong,G., Allen,K.M., Anisowicz,A., Morales,A.J.,
            Lomedico,P.T., Recker,S.M., Van Eerdewegh,P., Recker,R.R. and
            Johnson,M.L.
  TITLE     A mutation in the LDL receptor-related protein 5 gene results in
            the autosomal dominant high-bone-mass trait
  JOURNAL   Am. J. Hum. Genet. 70 (1), 11-19 (2002)
   PUBMED   11741193
  REMARK    GeneRIF: mutation results in an autosomal dominant high-bone-mass
            trait
REFERENCE   19  (bases 1 to 5100)
  AUTHORS   Gong,Y., Slee,R.B., Fukai,N., Rawadi,G., Roman-Roman,S.,
            Reginato,A.M., Wang,H., Cundy,T., Glorieux,F.H., Lev,D.,
            Zacharin,M., Oexle,K., Marcelino,J., Suwairi,W., Heeger,S.,
            Sabatakos,G., Apte,S., Adkins,W.N., Allgrove,J.,
            Arslan-Kirchner,M., Batch,J.A., Beighton,P., Black,G.C.,
            Boles,R.G., Boon,L.M., Borrone,C., Brunner,H.G., Carle,G.F.,
            Dallapiccola,B., De Paepe,A., Floege,B., Halfhide,M.L., Hall,B.,
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            Keppler-Noreuil,K., Kohlschuetter,A., LaCombe,D., Lambert,M.,
            Lemyre,E., Letteboer,T., Peltonen,L., Ramesar,R.S., Romanengo,M.,
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            Superti-Furga,A., Swoboda,W., van den Boogaard,M.J., Van Hul,W.,
            Vikkula,M., Votruba,M., Zabel,B., Garcia,T., Baron,R., Olsen,B.R.
            and Warman,M.L.
  CONSRTM   Osteoporosis-Pseudoglioma Syndrome Collaborative Group
  TITLE     LDL receptor-related protein 5 (LRP5) affects bone accrual and eye
            development
  JOURNAL   Cell 107 (4), 513-523 (2001)
   PUBMED   11719191
REFERENCE   20  (bases 1 to 5100)
  AUTHORS   Zorn,A.M.
  TITLE     Wnt signalling: antagonistic Dickkopfs
  JOURNAL   Curr. Biol. 11 (15), R592-R595 (2001)
   PUBMED   11516963
REFERENCE   21  (bases 1 to 5100)
  AUTHORS   Mao,J., Wang,J., Liu,B., Pan,W., Farr,G.H. III, Flynn,C., Yuan,H.,
            Takada,S., Kimelman,D., Li,L. and Wu,D.
  TITLE     Low-density lipoprotein receptor-related protein-5 binds to Axin
            and regulates the canonical Wnt signaling pathway
  JOURNAL   Mol. Cell 7 (4), 801-809 (2001)
   PUBMED   11336703
REFERENCE   22  (bases 1 to 5100)
  AUTHORS   Chen,D., Lathrop,W. and Dong,Y.
  TITLE     Molecular cloning of mouse Lrp7(Lr3) cDNA and chromosomal mapping
            of orthologous genes in mouse and human
  JOURNAL   Genomics 55 (3), 314-321 (1999)
   PUBMED   10049586
REFERENCE   23  (bases 1 to 5100)
  AUTHORS   Dong,Y., Lathrop,W., Weaver,D., Qiu,Q., Cini,J., Bertolini,D. and
            Chen,D.
  TITLE     Molecular cloning and characterization of LR3, a novel LDL receptor
            family protein with mitogenic activity
  JOURNAL   Biochem. Biophys. Res. Commun. 251 (3), 784-790 (1998)
   PUBMED   9790987
REFERENCE   24  (bases 1 to 5100)
  AUTHORS   Hey,P.J., Twells,R.C., Phillips,M.S., Yusuke,Nakagawa., Brown,S.D.,
            Kawaguchi,Y., Cox,R., Guochun,Xie., Dugan,V., Hammond,H.,
            Metzker,M.L., Todd,J.A. and Hess,J.F.
  TITLE     Cloning of a novel member of the low-density lipoprotein receptor
            family
  JOURNAL   Gene 216 (1), 103-111 (1998)
   PUBMED   9714764
REFERENCE   25 (bases 1 to 5100)
  AUTHORS   Johnson,M.L., Gong,G., Kimberling,W., Recker,S.M., Kimmel,D.B. and
            Recker,R.B.
  TITLE     Linkage of a gene causing high bone mass to human chromosome 11
            (11q12-13)
  JOURNAL   Am. J. Hum. Genet. 60 (6), 1326-1332 (1997)
   PUBMED   9199553
REFERENCE   26 (bases 1 to 5100)
  AUTHORS   Gong,Y., Vikkula,M., Boon,L., Liu,J., Beighton,P., Ramesar,R.,
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            and Warman,M.L.
  TITLE     Osteoporosis-pseudoglioma syndrome, a disorder affecting skeletal
            strength and vision, is assigned to chromosome region 11q12-13
  JOURNAL   Am. J. Hum. Genet. 59 (1), 146-151 (1996)
   PUBMED   8659519
REFERENCE   27 (bases 1 to 5100)
  AUTHORS   Godyna,S., Liau,G., Popa,I., Stefansson,S. and Argraves,W.S.
  TITLE     Identification of the low density lipoprotein receptor-related
            protein (LRP) as an endocytic receptor for thrombospondin-1
  JOURNAL   J. Cell Biol. 129 (5), 1403-1410 (1995)
   PUBMED   7775583
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does not encompass base 560

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>☐gi|55636538|ref|XM_508605.1| G PREDICTED: Pan troglodytes similar to LDL receptor mem
(LOC451380), mRNA
Length=6652

 Score = 42.1 bits (21),  Expect = 0.021
 Identities = 24/25 (96%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1      TCCTCCAGGTCGATGGTCAGCCCAT  25
              |||||||||||||||||||| ||||
Sbjct  1133   TCCTCCAGGTCGATGGTCAGTCCAT  1109


>☐gi|71052167|gb|BC098397.1|  Homo sapiens cDNA clone IMAGE:30527697
Length=5126

 Score = 42.1 bits (21),  Expect = 0.021
 Identities = 24/25 (96%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1     TCCTCCAGGTCGATGGTCAGCCCAT  25
             |||||||||||||||||||| ||||
Sbjct  648   TCCTCCAGGTCGATGGTCAGTCCAT  624


>☐gi|14028616|gb|AF283320.1|AF283320S1 D Homo sapiens low density lipoprotein receptor-
5 (LRP5) gene, exons 1 through 9
Length=122056

 Score = 42.1 bits (21),  Expect = 0.021
 Identities = 24/25 (96%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1       TCCTCCAGGTCGATGGTCAGCCCAT  25
               |||||||||||||||||||| ||||
Sbjct  69653   TCCTCCAGGTCGATGGTCAGTCCAT  69629


>☐gi|3831747|gb|AF077820.1|AF077820 U E G Homo sapiens LDL receptor member LR3 mRNA, co
Length=5135

 Score = 42.1 bits (21),  Expect = 0.021
 Strand=Plus/Minus

Query  1    AGTACAGCTTCTTGCCAACCCAGTC  25
            |||||||||||||||||||||||||
Sbjct  479  AGTACAGCTTCTTGCCAACCCAGTC  455


>☐gi|15030227|gb|BC011374.1| U E G Mus musculus low density lipoprotein receptor-relatec
5, mRNA (cDNA clone MGC:11611 IMAGE:3154246), complete cds
Length=5166

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1    AGTACAGCTTCTTGCCAACCCAGTC  25
            |||||||||||||||||||||||||
Sbjct  471  AGTACAGCTTCTTGCCAACCCAGTC  447


>☐gi|3831749|gb|AF077847.1|AF077847 U E G Mus musculus LDL receptor member LR3 mRNA, cor
Length=5043

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1    AGTACAGCTTCTTGCCAACCCAGTC  25
            |||||||||||||||||||||||||
Sbjct  367  AGTACAGCTTCTTGCCAACCCAGTC  343


>☐gi|3641528|gb|AF064984.1|AF064984 U E G Mus musculus low-density lipoprotein receptor-
5 (Lrp5) mRNA, complete cds
Length=5119

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1    AGTACAGCTTCTTGCCAACCCAGTC  25
            |||||||||||||||||||||||||
Sbjct  479  AGTACAGCTTCTTGCCAACCCAGTC  455


>☐gi|4505018|ref|NM_002335.1| U E G Homo sapiens low density lipoprotein receptor-relate
5 (LRP5), mRNA
Length=5100

 Score = 42.1 bits (21),  Expect = 0.021
 Identities = 24/25 (96%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1    AGTACAGCTTCTTGCCAACCCAGTC  25
            |||||||||||||||| ||||||||
Sbjct  418  AGTACAGCTTCTTGCCCACCCAGTC  394
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does not encompass base 560

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>☐gi|55636538|ref|XM_508605.1| G PREDICTED: Pan troglodytes similar to LDL receptor memk
(LOC451380), mRNA
Length=6652
```

```
gi|6678715|ref|NM_008513.1|  Mus musculus low density lipoprot...  34.2   5.0   UEG
gi|15030227|gb|BC011374.1|  Mus musculus low density lipoprote...  34.2   5.0   UEG
gi|3831749|gb|AF077847.1|AF077847  Mus musculus LDL receptor memb  34.2   5.0   UEG
gi|28610524|emb|AL840631.9|  Zebrafish DNA sequence from clone...  34.2   5.0
gi|3641528|gb|AF064984.1|AF064984  Mus musculus low-density li...  34.2   5.0   UEG
gi|24394981|emb|AL731703.16|  Mouse DNA sequence from clone RP...  34.2   5.0
gi|81778297|gb|AC165249.2|  Mus musculus BAC clone RP24-157P13...  34.2   5.0
gi|6683557|dbj|AB024565.1|  Mus musculus mRNA for heparan sulf...  34.2   5.0   UEG
gi|21425240|emb|AL671918.5|  Mouse DNA sequence from clone RP2...  34.2   5.0
```

Alignments

Get selected sequences | Select all | Deselect all

```
>☐gi|4505018|ref|NM_002335.1| UEG Homo sapiens low density lipoprotein receptor-relate
5 (LRP5), mRNA
Length=5100

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1     GTCTGAGTCCGAGTTCAAATCCAGG  25
             |||||||||||||||||||||||||
Sbjct  4761  GTCTGAGTCCGAGTTCAAATCCAGG  4737
```

does not encompass base 560

```
>☐gi|55636538|ref|XM_508605.1| G PREDICTED: Pan troglodytes similar to LDL receptor memk
(LOC451380), mRNA
Length=6652

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1     GTCTGAGTCCGAGTTCAAATCCAGG  25
             |||||||||||||||||||||||||
Sbjct  6327  GTCTGAGTCCGAGTTCAAATCCAGG  6303


>☐gi|71052167|gb|BC098397.1|  Homo sapiens cDNA clone IMAGE:30527697
Length=5126

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
 Strand=Plus/Minus

Query  1     GTCTGAGTCCGAGTTCAAATCCAGG  25
             |||||||||||||||||||||||||
Sbjct  4766  GTCTGAGTCCGAGTTCAAATCCAGG  4742


>☐gi|14028617|gb|AF283321.1|AF283320S2 ED Homo sapiens low density lipoprotein recepto
5 (LRP5) gene, exons 10 through 23 and complete cds
Length=64475

 Score = 50.1 bits (25),  Expect = 8e-05
 Identities = 25/25 (100%), Gaps = 0/25 (0%)
```